Disease	Defect	Symptoms	Treatment
Cystinuria	1.) Defect in the cationic amino acids (ornithine, lysine, and arginine) and cystine 2.) Type A - caused by mutation in the heavy chain Type B in light chain Type A/B is mutation in both SLC3A1 and SLC7A1 is full blown Cystinuria	Excrete large amounts of cysteine in the urine. Positive in the cyanide-nitroprusside test.	1.) adaquate hydration 2.) Keeping urine slightly alkaline 3.) Reduction in dietary intake of cystein and methionine. 4.) treatment with agents (penicellamine and alpha-mercaptopropionyglycine (Thiopronin))
Hartnup Disorder	1.) Defects in the transporter for neutral amino acids 2.) Transporter is Na+ dependent 3.) mostly in small intestine and kidney but also other tissues like the brain 5.) Interaction with Collectrin and ACE2	1.) Marked by poor tryptophan uptake. 2.) rash with light sensitivity, psychosis, cerebellar ataxia 3.) Tryptophan is a precursor for nicatinamide and also serotonin 4.) Elevated neutral amino acids in the urine.	1.) Treatment with niacin
Lysinuric protein Intolerance	1.) Autosommal recessive defect in the basolateral transporter (LAT1) for cationic amino acids 2.) Defective uptake of lysine, argininem and ornithine in small intestine and kidney	1.) Increase excretion of cationic aa. 2.) Derangement of urea cycle because low ornithine. 3.) Neurological problems because increased ammonia 4,) LAT II in lower conc. cant overcome. 5.) Aversion to protein 6.) Growth retard. enlarged liver and spleen, muscular hypotonia, osteoperosis	1.) Sodium benzoate and phenylbutarate to promote excretion of nitrogen 2.) Oral citrulline - precursor to arginine and ornithine. 3.) Limited protein intake
Maple Syrup Urine Disease (MSUD)	1.) branched chain keto acid dehydrogenase 2.) catalyzes the conversion of branched chain keto acids to Coa derivatives	1.) Leucine, Isoleucine and Valine in blood (alloisoleucine is diagnostic) 2.) burnt sugar of maple smell in the urine. 3.) Lethargy and no interest in feeding 4.) BCKD activity lowered. 5.) weight loss and neurological deterioration	1.) Must remove toxic metabolites (especially leucine) early on by blood transfusion or hemodialysis 2.) Thiamine may improve tolerance of BCAA in some patients 3.) low protein diet

Disease	Defect	Symptoms	Treatment
Isovaleric Acidemia	1.) Isovaleryl CoA dehydrogenase 2.) catalyzes the conversion of isovaleryl CoA to Beta methylcrotonyl CoA	1.) First Type- within two weeks - vomiting, dehydration, listlessness, acidosis. 2.) Type 2 - chronic and intermittent form - vomiting, lethargy to come, anion gap, dirty sock odor, metabolic acidosis, ketonuria, 3.) exacerbated by stress	1.) Dietary restriction of proteins 2.) diet with mixture of amino acids free of leucine. 3.) oral glycine and intravenous carnitine (react with isovaleryl-CoA to get non toxic forms)
Methymalonic Acidemia (Aciduria) **Propionic Acidemia (Aciduria)**	1.) Defects in either Propionyl CoA carboxylase or Methylmalonyl CoA mutase a.) Propionyl CoA carboxylase- catalyzes prop coA to methylmalonyl CoA (**PCC is biotin dependent!)** b.) Methylmalonyl coA mutase catalyzes conversion to Succinyl CoA	1.) Accumulation of toxic metabolites (elevated propionic acid in the blood and urine 2.) impaired mitochondrial energy production (propionyl-CoA inhibits glycolysis and urea cycle. 3.) Elevated serum ammonia. 4.) GI and neurological signs patients will go into coma 5.) accumulation of metabolites in brain; kidney failure; cardiac myopathy	Prognosis grim
Nonketotic Hyperglycinemia	1.) Defects in the glycine cleavage system (glycine synthase) 2.) Remember system found in liver kidney and brain but not heart. 3.) Contains P-, H-, T-, and L-protein.	1.) 80% mutation in P protein, 20% in T-prot. 2.) High levels of glycine in the plasma and cerebrospinal fluid 3.) neurological signs: lethargy vomiting, convulstions, loss of primitive reflexes 4.) Hiccups! 5.) High glycine affects Glycine receptors in inhibitory neurons, NMDA gluatmate receptors, and abnormal metabolism of H4folate.	1.) Reduce activity of the NMDA with Diazapam and Ketamine and dextromethrophan 2.) Promote excretion of glycine (Sodium Benzoate) 3.) Controlling seizures - (Phenobarbital)

Hyperammonenia Type 1	Hyperammonenia Type II	Argininosuccinate synthetase deficiency	Argininosuccinate lyase deficiency	Arginase deficiency
a.) Carbamoyl phosphate synthetase I (CPI) deficiency Treatment with arginine stimulates CPI. b.) N-acetylglutamate synthase deficiency – treatment with carbamoyl glutamate activates CPI.	Ornithine transcarbamylase deficiency – Elevated blood ammonia, amino acids, orotic acid, glutamine. – X-linked	Argininosuccinate synthetase deficiency – Citrulline & ammonia elevated in plasma, CSF and urine. – Arginine treatment enhances citrulline excretion.	-Argininosuccinate and ammonia in plasma, CSF and urine – Treated with arginine	– Arginine and ammonia elevated in plasma, CSF and urine; arginine, lysine and ornithine in urine. – Treatment: diet of essential amino acids, minus arginine. Low protein diet reduces plasma ammonia.